

## **PARENT INFORMATION for Initial INCONCLUSIVE newborn screening results for CONGENITAL ADRENAL HYPERPLASIA (CAH)**



When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually can not be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were "inconclusive" for a disorder called congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia). It is important to remember this is only a screening test. A new specimen will be collected from your baby and tested to help diagnose your baby, or to rule out this disorder.

### **? What does an "inconclusive" screen mean?**

A small percent of all babies screened will be identified as "inconclusive" on screening, and most are later found to not have the disorder. A repeat screening test should be done to determine if your child has this disorder, or to rule it out. Keep in mind that the new test could show that your baby doesn't have CAH.

### **👉 What is the next step?**

Be sure to follow your baby's doctor's instructions for getting a new specimen for further testing as soon as you receive them. In the meantime, before the results of the additional test(s) are available, your doctor will talk with you about your baby's current health. CAH is a rare disease involving the endocrine system, so your baby's doctor might consult with or refer you to a pediatric endocrinologist.

### **? What exactly is CAH?**

CAH is rare disease caused by a missing enzyme. When this enzyme is missing, the adrenal glands produce too much of some hormones and not enough

of others. The salt-retaining hormone is one that is under-produced. Therefore babies with this condition are at risk of a serious sudden "salt-wasting" episode that can be fatal. A baby with CAH may appear normal at birth. Be sure to call your baby's doctor if any of these symptoms appear: a lot of vomiting, poor weight gain or if your baby looks ill.

There can also be other forms of CAH with less serious but still significant consequences. Children will usually develop normally if treatment begins in the first weeks of life. Finding out early if your baby has CAH, means you and your baby's doctor can take steps to prevent these effects.

### **👣 What can I do to prevent these effects?**

The most important thing that you can do is to be sure your baby goes in for a new specimen to be collected and tested as soon as possible. If it turns out your baby does have CAH, there are very effective treatments available. The pediatric endocrinologists will be able to help you manage this through monitoring the blood levels and adjusting the medication to help normalize hormone levels.

You have probably already done this, but now is also a good time to be sure your baby is covered by your health insurance provider.

NO treatment should be done unless a physician recommends it. Therapy should NOT be started before collecting the new specimen.

Remember, this could be a false alarm, but if it turns out your baby does have CAH, the treatment is very effective.

For more information talk with your baby's doctor, or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Endocrinologists available in Nebraska. A good web-site for more information can be found at [www.caresfoundation.org](http://www.caresfoundation.org) or [www.magicfoundation.org](http://www.magicfoundation.org).